

CLAIMS

1. A purified preparation of antibodies which specifically bind to a mutant human surfactant protein C comprising an amino acid alteration due to the presence of a single nucleotide polymorphism (SNP) in a gene encoding the mutant surfactant protein C, wherein the SNP is associated with interstitial lung disease, wherein the antibodies do not bind to a wild-type human surfactant protein C.
2. The preparation of claim 1 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.
3. The preparation of claim 1 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.
4. The preparation of claim 3 wherein the nucleotide at position 460 +1 is adenine.
5. The preparation of claim 3 wherein the nucleotide at position 460 +1 is thymidine.
6. The preparation of claim 1 wherein the antibodies are polyclonal antibodies.
7. The preparation of claim 1 wherein the antibodies are monoclonal antibodies.
8. The preparation of claim 1 wherein the antibodies are Fab, F(ab')₂, or Fv fragments.

9. A single-stranded polynucleotide comprising 12 contiguous nucleotides of a mutant allele of a human surfactant protein C gene, wherein the 12 contiguous nucleotides comprise a SNP associated with interstitial lung disease.

10. The single-stranded polynucleotide of claim 9 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 49, 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.

11. The single-stranded polynucleotide of claim 9 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.

12. The single-stranded polynucleotide of claim 11 wherein the nucleotide at position 460 +1 is adenine.

13. The single-stranded polynucleotide of claim 11 wherein the nucleotide at position 460 +1 is thymidine.

14. The single-stranded polynucleotide of claim 9 which comprises a detectable label.

15. The single-stranded polynucleotide of claim 9 wherein the SNP is at either the 3' or the 5' end of the polynucleotide.

16. The single-stranded polynucleotide of claim 9 which is bound to a solid support.

17. A kit, comprising:

a reagent for detecting a SNP in a mutant allele of a human surfactant protein C gene, wherein the SNP is associated with interstitial lung disease; and instructions for a method of detecting the SNP.

18. The kit of claim 17 wherein the reagent is an antibody which specifically binds to a mutant human surfactant protein C comprising an amino acid alteration due to the presence of the SNP, wherein the antibody does not bind to a wild-type human surfactant protein C.

19. The kit of claim 17 wherein the reagent is a single-stranded polynucleotide comprising 12 contiguous nucleotides of the mutant allele, wherein the 12 contiguous nucleotides comprise the SNP or the complement of the SNP.

20. The kit of claim 17 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 49, 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.

21. The kit of claim 17 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.

22. The kit of claim 21 wherein the nucleotide at position 460 +1 is adenine.

23. The kit of claim 21 wherein the nucleotide at position 460 +1 is thymidine.

24. A method of identifying an individual as predisposed to developing interstitial lung disease associated with a defect in surfactant protein C, comprising the steps of:

assaying a biological sample obtained from the individual to determine if an allele of a surfactant protein C gene comprises a SNP associated with interstitial lung disease; and

identifying the individual as predisposed to developing the interstitial lung disease if the allele comprises the SNP.

25. The method of claim 24 wherein the biological sample is lung tissue.

26. The method of claim 24 wherein the biological sample is bronchoalveolar lavage fluid.

27. The method of claim 24 wherein the biological sample is blood.

28. The method of claim 24 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 49, 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.

29. The method of claim 24 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.

30. The method of claim 29 wherein the nucleotide at position 460 +1 is adenine.

31. The method of claim 29 wherein the nucleotide at position 460 +1 is thymidine.

32. The method of claim 24 wherein surfactant protein C in the biological sample is assayed to detect an amino acid alteration due to the presence of the SNP.

33. The method of claim 32 wherein the surfactant protein C is assayed using an antibody which specifically binds to a mutant surfactant protein C comprising the amino acid alteration, wherein the antibody does not bind to a wild-type surfactant protein C.

34. The method of claim 25 wherein nucleic acid is assayed to detect the SNP.

35. The method of claim 34 wherein the nucleic acid is assayed using a single-stranded polynucleotide comprising 12 contiguous nucleotides of a mutant allele of the surfactant protein C gene, wherein the 12 contiguous nucleotides comprise the SNP or the complement of the SNP.

36. The method of claim 25 wherein the interstitial lung disease is desquamative interstitial pneumonitis.

37. A method of diagnosing interstitial lung disease associated with a defect in surfactant protein C, comprising the steps of:

assaying a biological sample obtained from an individual to determine if an allele of a surfactant protein C gene comprises a SNP associated with interstitial lung disease; and

identifying the individual as having the interstitial lung disease if the allele comprises the SNP.

38. The method of claim 37 wherein the biological sample is lung tissue.

39. The method of claim 37 wherein the biological sample is bronchoalveolar lavage fluid.

40. The method of claim 37 wherein the biological sample is blood.

41. The method of claim 37 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 49, 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.

42. The method of claim 37 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.

43. The method of claim 42 wherein the nucleotide at position 460 +1 is adenine.

44. The method of claim 42 wherein the nucleotide at position 460 +1 is thymidine.

45. The method of claim 37 wherein surfactant protein C in the biological sample is assayed to detect an amino acid alteration due to the presence of the SNP.

46. The method of claim 45 wherein the surfactant protein C is assayed using an antibody which specifically binds to a mutant surfactant protein C comprising the amino acid alteration, wherein the antibody does not bind to a wild-type surfactant protein C.

47. The method of claim 37 wherein nucleic acid is assayed to detect the SNP.

48. The method of claim 47 wherein the nucleic acid is assayed using a single-stranded polynucleotide comprising 12 contiguous nucleotides of a mutant allele of the surfactant protein C gene, wherein the 12 contiguous nucleotides comprise the SNP or the complement of the SNP.

49. A method of determining whether an individual having interstitial lung disease is likely to respond to a therapeutic intervention, comprising the steps of:

assaying a biological sample obtained from the individual to determine whether both alleles of the individual's surfactant protein C gene comprise a SNP associated with interstitial lung disease; and

identifying the individual as likely to respond to the therapeutic intervention if neither allele comprises the SNP.

50. The method of claim 49 wherein the biological sample is lung tissue.

51. The method of claim 49 wherein the biological sample is bronchoalveolar lavage fluid.

52. The method of claim 49 wherein the biological sample is blood.

53. The method of claim 49 wherein the SNP is located at a nucleotide position of SEQ ID NO:1 selected from the group consisting of nucleotide positions 49, 114, 219, 243, 246, 324, 332, 335, 359, 369, 402, 443-445, the intronic nucleotide immediately 3' of nucleotide 460 (460 +1), 521-523, 585, and 588.

54. The method of claim 49 wherein the SNP is located at nucleotide position 460 +1 of SEQ ID NO:1.

55. The method of claim 54 wherein the nucleotide at position 460 +1 is adenine.

56. The method of claim 54 wherein the nucleotide at position 460 +1 is thymidine.

57. The method of claim 49 wherein the therapeutic intervention is administration of a glucocorticoid.

58. The method of claim 49 wherein the therapeutic intervention is administration of chloroquine.